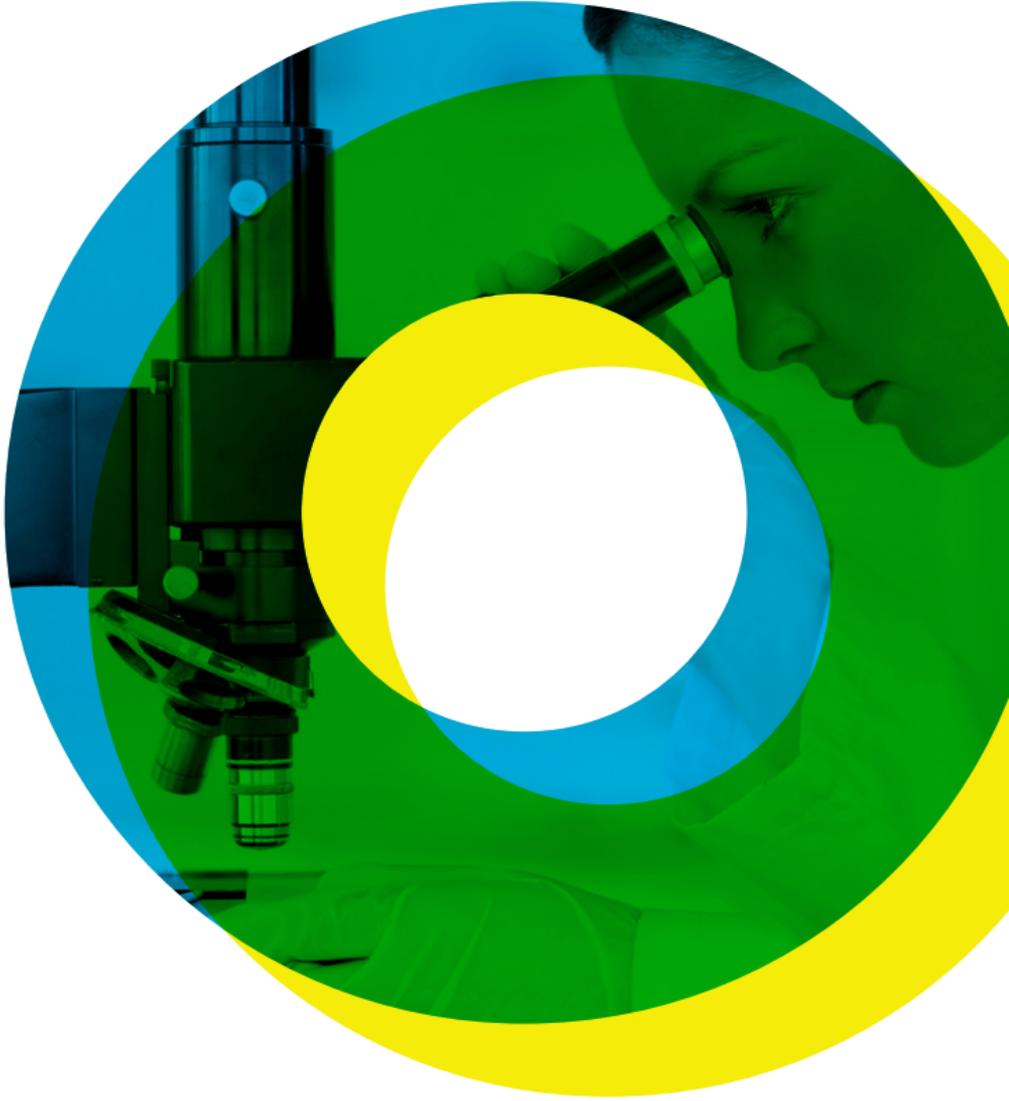


LABORS.AT



GENEVIEW:  
MYOCARDIAL INFARCTION

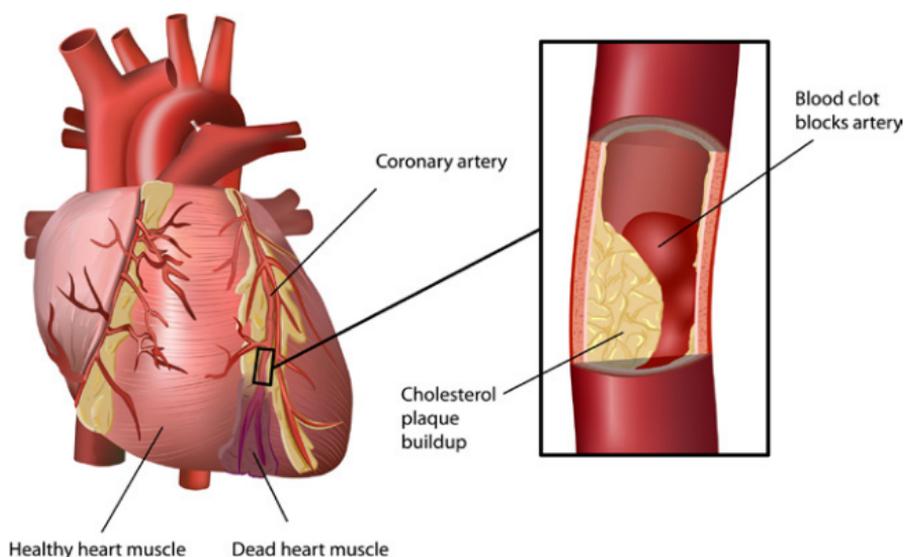
CAUSES | RISK FACTORS | PREVENTION

## WHAT IS CORONARY HEART DISEASE? WHAT IS A HEART ATTACK?

All organs in the human body are supplied with nutrients and oxygen by the blood, which is transported to the organs via special blood vessels (arteries).

If cholesterol, fat, and calcium accumulate for years in the vascular wall (arterial sclerosis, vascular calcification), the vascular wall thickens and the internal diameter of the affected vessel decreases. The location of wall thickening is called plaque. If the coronary arteries are affected — those are the arteries that supply blood to the heart — then the patient suffers from coronary heart disease.

Narrowing of the coronary arteries leads to decreased coronary blood flow and chest pains (angina pectoris), especially during physical exertion.



If a plaque breaks up, a blood clot forms at the narrowed portion of the artery, which occludes the artery. This cuts off a portion of the heart muscle from oxygen supply, and this portion dies. A myocardial infarction has occurred.

The process described above can also occur in other regions of the body. If a blood vessel (whose task is to supply a certain region of the brain) becomes blocked, this portion of the brain dies, which is known as a cerebral infarction or stroke.

Heart attack is the second most common cause of death in Austria. Circa 50 % of patients with heart attacks die as a result. Thus, it is crucial to explore all ways of preventing heart attacks.

## **HOW CAN CORONARY HEART DISEASE BE DIAGNOSED?**

There are various examination procedures that can be used to diagnose coronary heart disease; e.g. detection of reduced cardiac circulation under stress or detecting calcium deposits in the coronary arterial walls.

A coronary angiogram will be performed if the existence of coronary heart disease is reasonably suspected. This is an x-ray examination in which contrast medium is injected into the coronary arteries. This renders the walls of the contrast-filled arteries easier to evaluate.

## ARE THERE TREATMENT OPTIONS FOR EXISTING CORONARY HEART DISEASE?

Treatment for coronary heart disease consists of carefully avoiding risk factors and of intensive medication and surgical treatment, if necessary. The goal of treatment is stabilisation of, or, in early stages of the disease, reversal of changes to the coronary arterial walls.

If the arteries are severely affected, if complications arise, or if a heart attack has already occurred, then often surgical measures must be taken to improve coronary circulation, in addition to avoiding risk factors and undergoing medication treatment.

In these cases, a cardiac catheter is used to insert a vessel-implant (stents) into the affected coronary artery in order to stretch it and thus achieve sufficient blood flow. If this is not possible, then blood vessels are taken from another area of the patient's body and attached to bypass the narrowed arteries (bypass operation).

**Coronary heart disease is a life-threatening illness. Everyone should know his or her personal risk profile. People at risk with an unfavourable constellation of risk factors should do everything possible to prevent the onset of coronary heart disease and to delay its progress.**

## **WHICH RISK FACTORS FOR DEVELOPING CORONARY HEART DISEASE AND ITS CONSEQUENCE — MYOCARDIAL INFARCTION — ARE CURRENTLY KNOWN?**

If someone suffers from myocardial infarction, it is not merely an act of fate.

A number of risk factors for myocardial infarction have been defined:

- \_ Age**
- \_ Male sex**
- \_ Family history of heart attack**
- \_ Elevated cholesterol levels**
- \_ Elevated blood sugar levels**
- \_ Elevated blood pressure**
- \_ Smoking**

These risk factors promote the onset of coronary heart disease as described above (thickening of arterial walls due to cholesterol, fat, and calcium).

If nothing is done to prevent the disease or its progress, the consequences can be fatal — as in the case of myocardial infarction.

## **ARE THERE PREVENTATIVE MEASURES AGAINST DEVELOPING CORONARY HEART DISEASE AND ITS CONSEQUENCE, NAMELY MYOCARDIAL INFARCTION?**

Various measures are known to counteract the risk factors, leading to a reduction in overall risk.

The following measures have been shown to reduce the risk of myocardial infarction:

### **Healthy diet**



### **Losing excess weight, if overweight**



## Physical activity



## Avoiding nicotine



## Avoiding stress



## **Diet / medication against elevated blood lipid or blood sugar values**



## **Medication against elevated blood pressure**



Therefore, it makes sense to know your own risk situation and work with your treating physician, if necessary, to establish measures to lower your personal risk.

## IS THERE NEW KNOWLEDGE ABOUT MYOCARDIAL INFARCTION RISK FACTORS?

It has been known for a long time that the risk of myocardial infarction has a hereditary component. This is probably because the prevalence of some known risk factors is inherited. The rapid development of new molecular genetic examination techniques has led to the discovery of numerous previously unknown variants of human genes associated with an increased risk of heart myocardial infarction. Some of them are not associated with known cardiovascular risk factors.

These variants are certain changes in the DNA code of genes. The chromosomes, found in the nucleus of each human cell, contain the entire blueprint in the form of genes. In different cell types (skin cells, muscle cells, brain cells, etc.) only the particular part of the blueprint needed by those specific cells is activated.

The blueprint consists of deoxyribonucleic acid (DNA) whose regular construction is necessary for healthy functioning of individual cells.

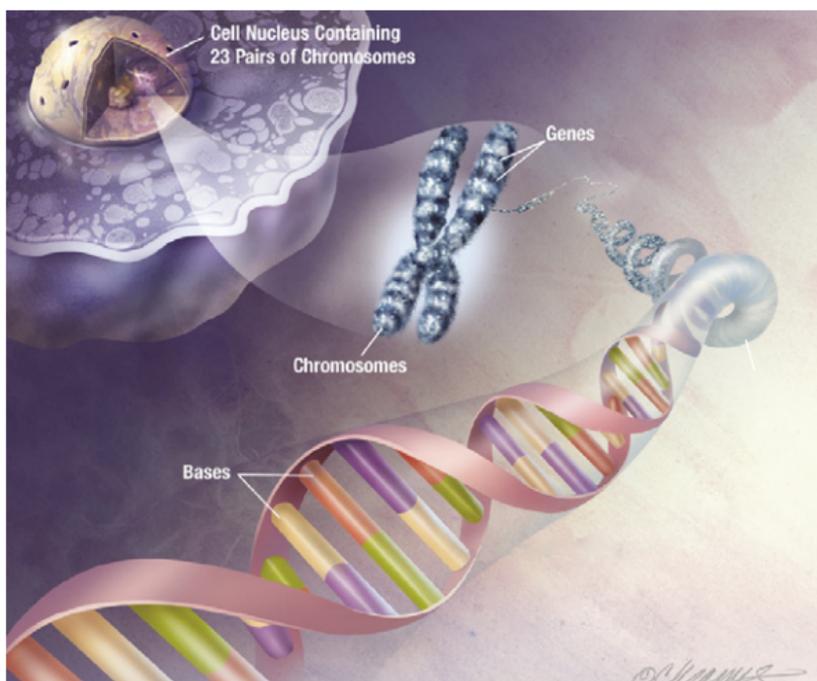


Image: National Institutes of Health

healthy DNA



SNP

modified DNA



Human genes are built from so-called nucleotides (C, T, A, G). Exchanging a single nucleotide is called single nucleotide polymorphism (SNP), and can be the cause of disease or of an increased risk for certain diseases

Errors in DNA assembly, so called mutations due to incorrect insertion of base-pairs / nucleotides can lead to malfunctioning of cells and to the development of diseases. DNA mutations can also be caused by external influences (toxins, UV exposure, etc.), or they may be inherited. If only a single nucleotide is changed at a certain point in the DNA sequence, this is described as single nucleotide polymorphism (SNP).

## IS IT POSSIBLE TO CALCULATE MY PERSONAL RISK OF SUFFERING FROM MYOCARDIAL INFARCTION?

For many years, factors such as age, sex, cholesterol and blood sugar levels, blood pressure, and smoking cigarettes have been used to calculate personal risk of myocardial infarction. Every person can also be assigned a certain level of risk according to the internationally accepted Framingham calculation method.

### **Framingham risk levels:**

\_ **Low risk < 10%**

\_ **Average risk 10–20%**

\_ **High risk > 20%**

**Within the high-risk group, 20 out of every 100 people will suffer from myocardial infarction within the next 10 years.**

The treatment chosen by the treating physician is determined by the risk level. There are internationally accepted guidelines for treatment.

## IS THERE A RISK CALCULATION METHOD FOR MYOCARDIAL INFARCTION WHICH ALSO INCLUDES GENETIC FACTORS?

The Framingham calculation method is a proven method of risk calculation, though it has the disadvantage of recognizing only the risk factors that have been known for many years, and not the newly discovered genetic abnormalities associated with the risk of disease.

Labors.at has thus developed a risk calculation method (based on the Framingham risk score) taking into consideration newly discovered genetic risk factors. This allows more precise assessment of risk and thus improved individual treatment suggestions.

This is of special importance if patients, in consideration of previously known risk factors, are assigned to a low

level of risk and receive treatment inadequate for their actual level of risk (or perhaps no treatment, at all).

**A diagnosis of increased genetic risk of myocardial infarction with the GeneView myocardial infarction risk assessment should be especially motivating to pay close attention to one's health and gives your physician the tool to develop an individualised prevention strategy.**

In any case, the results of the GeneView myocardial infarction risk assessment should be discussed with a specialist in cardiovascular disease.

### **Case Report:**

**A 60 year old male presents with a systolic blood pressure of 145/80 mmHg as a non-smoker, non-diabetic with a total cholesterol of 220mg/dl and a HDL cholesterol of 50 mg/dl. A conventional Framingham risk score (without consideration of the genetic background) yields a personal ten year risk of 16.6% of suffering a heart attack or any major cardiovascular event. This is generally considered as medium risk (10-20%). After inclusion of his personal genetic background (shown in the table as variant 1), his personal risk would slightly decrease to 13.6%. In contrast a genetic background as described in variant 2 would result in an elevated ten year risk of 24.2%, considered as a high cardiovascular risk. This patient might benefit from personalized prevention strategies.**

Genetic Variant (SNP)	Number of risk-alleles* for each variant	
	Variant 1	Variant 2
17465637	0	1
6725887	0	0
9818870	1	1
12526453	1	1
1333049	0	2
1746048	2	2
9982601	1	1
10455872	0	2
<b>Cardiovascular risk</b>	<b>Variant 1</b>	<b>Variant 2</b>
including genetic risk factors	13.6 %	24.2 %
without considering genetic risk factors	16.6 %	

\*from each parent one allele is inherited, either with or without elevated risk. Thus each person can have nil, one or two risk-alleles for each genetic variant.

In the example described above, determining genetic risk factors by means of the GeneView myocardial infarction risk assessment leads the patient to be assigned to a different group, affecting the course of treatment.

The GeneView myocardial infarction risk assessment guarantees that your personal assessment of the myocardial infarction risk and any treatment measures chosen take into account the most up-to-date scientific knowledge.

## WHERE AND HOW IS THE GENEVIEW MYOCARDIAL INFARCTION RISK ASSESSMENT PERFORMED?

The GeneView myocardial infarction risk assessment can be performed with a simple blood sample. You can have a sample drawn at Labors.at (appointment and information under +43 (0)1 260 53-100), or you can have your treating physician send the sample to our laboratory.

A consultation always is made at the beginning of your risk assessment. This can be done by a Labors.at specialist in laboratory medicine or by your treating physician. This consultation will be used to obtain any personal data necessary for performing the assessment. During the consultation, you will be provided with information about the price, nature, scope, and significance of the planned examination. In addition, any possible implications which may be drawn from the assessment results will be discussed with you.

You may choose whether you would prefer to receive your results per post or if you would like to pick up your results personally.

It is highly recommended that you discuss these results with your treating physician (internal medicine/ cardiology/angiology or general medicine specialist, etc.).

If you decide to take measures to decrease your personal risk of myocardial infarction, Labors.at offers a follow-up examination. This follow-up examination will not re-determine your genetic markers, since these remain unchanged throughout your life. The new parameters affected by your risk-decreasing measures will be considered in the updated myocardial infarction risk assessment. This allows you to objectively evaluate any success of your efforts and the change in your risk situation.

The GeneView myocardial infarction risk assessment is a private service and is not covered by health insurance. You can find the current prices and additional informations on our website under [www.labors.at](http://www.labors.at) → Patienten → Informationen → Genuntersuchungen Patienteninformation → Genuntersuchung GeneView Herzinfarkttrisiko.

## ADDITIONAL INFORMATION

Please contact us for additional information:

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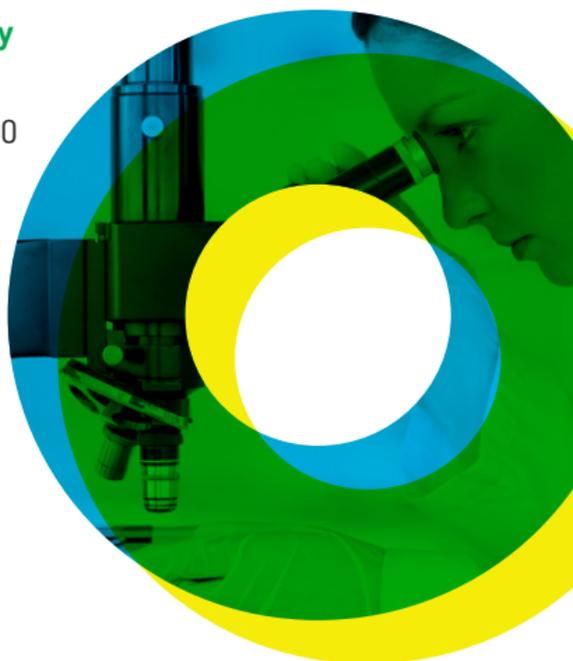
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